

Freidrich's Ataxia

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Friedreich's ataxia is an **inherited disease** that causes progressive damage to the nervous system resulting in symptoms ranging from **gait disturbance** and **speech problems** to **heart disease**. It is named after the physician Nikolaus Friedreich, who first described the condition in the 1860s. "Ataxia," which refers to coordination problems such as clumsy or awkward movements and unsteadiness, occurs in many different diseases and conditions. The ataxia of Friedreich's ataxia results from the **degeneration of nerve tissue** in the spinal cord and of nerves that control muscle movement in the arms and legs. The **spinal cord becomes thinner and nerve cells lose some of their myelin sheath** — the insular covering on all nerve cells that helps conduct nerve impulses.



Symptoms usually begin between the ages of 5 and 15 but, on rare occasions, can appear as early as 18 months or as late as 50 years of age. The first symptom to appear is usually **difficulty in walking, or gait ataxia**. The ataxia gradually worsens and slowly spreads to the arms and then the trunk. Over time, muscles begin to weaken and waste away, especially in the feet, lower legs, and hands and deformities develop. Other symptoms include loss of tendon reflexes, especially in the knees and ankles. There is often a gradual loss of sensation in the extremities, which may spread to other parts of the body. Dysarthria (**slowness and slurring of speech**) develops, and the person is easily fatigued. Rapid, rhythmic, **involuntary movements of the eye** (nystagmus) are common. Most people with Friedreich's ataxia develop scoliosis (a curving of the spine to one side), which, if severe, may impair breathing.

Other symptoms that may occur include chest pain, shortness of breath, and heart palpitations. These symptoms are the result of **various forms of heart disease that often accompany Friedreich's ataxia**. About 20 percent of people with Friedreich's ataxia develop carbohydrate intolerance and 10 percent develop diabetes mellitus. Some people lose hearing or eyesight.

The rate of progression varies from person to person. Generally, within 10 to 20 years after the appearance of the first symptoms, the person is confined to a wheelchair, and in later stages of the disease individuals become completely incapacitated. Life expectancy may be affected, and many people with Friedreich's ataxia die in adulthood from the associated heart disease, the most common cause of death. However, some people with less severe symptoms of Friedreich's ataxia live much longer, sometimes into their sixties or seventies.

As with many degenerative diseases of the nervous system, there is currently no cure or effective treatment for Friedreich's ataxia. However, **many of the symptoms and accompanying complications can be treated to help patients maintain optimal functioning as long as possible**. Diabetes, if present, can be treated with diet and medications such as insulin, and some of the heart problems can be treated with medication as well. Orthopedic problems such as foot deformities and scoliosis can be treated with braces or surgery. Physical therapy may prolong use of the arms and legs. Scientists hope that recent advances in understanding the genetics of Friedreich's ataxia may lead to breakthroughs in treatment.

Expert in Freidrich's Ataxia? DSES is always looking for volunteers to help teach or give presentations!!

Resources and Additional Information

National Institute of Neurological Disorders and Stroke; www.ninds.nih.gov

Photo: Freidrich's Ataxia Research Alliance; www.curefa.org